

Ectopia Cordis: A Case Report

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ABSTRACT

Pentalogy of Cantrell is a rare congenital syndrome which includes ectopia cordis, sternal, pericardial, abdominal wall defects (usually omphalocele), diaphragmatic defects and

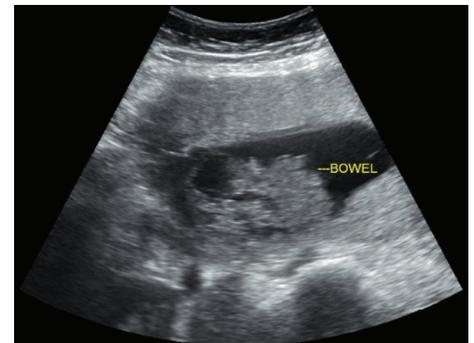
intracardiac abnormalities. We report a case with a rare combination of ectopia cordis omphalocele, dysplastic kidneys, megabladder and a malrotated pelvis in a 23-years old woman who had come for an antenatal checkup in her 25th week of gestation.

Keywords: Cardiac defect, Congenital anomaly, Pentalogy of Cantrell

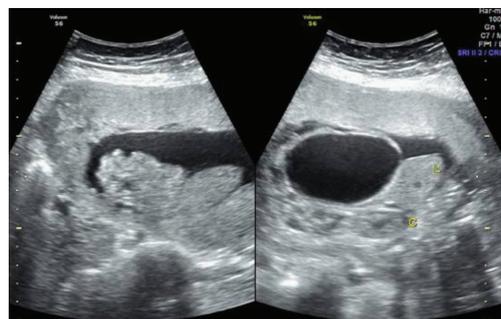
CASE REPORT

A 23-year-old pregnant woman with an obstetric history of G3P2A1 with 1 previous history of spontaneous abortion (cause could not be established) and one normal child. She had no family history of genetic disorders and no history of drug/ radiation exposure during the pregnancy.

It was her 25th week of pregnancy and she was referred for an anomaly ultrasound scan by her doctor. Ultrasound revealed decreased thoraco - abdominal measurement, fetal heart outside the thoracic cavity [Table/Fig-1,2] the bowel and liver were found outside the abdominal cavity covered with a thin membrane [Table/Fig-1,3-5]. Kidneys were dysplastic and the bladder was enlarged extending from the pelvis up



[Table/Fig-1]: Ultrasound image showing heart outside the thoracic cavity. **[Table/Fig-2]:** Color flow within the heart in color Doppler imaging. **[Table/Fig-3]:** Ultrasound image showing bowel out of the abdominal cavity.



[Table/Fig-4]: Ultrasound image floating bowel loops. **[Table/Fig-5]:** Ultrasound image showing bilateral kidney with few cyst (c), dilated bladder and floating liver. **[Table/Fig-6]:** Ultrasound image showing umbilical vessels and dilated bladder.

to the abdomen [Table/Fig-5,6]. The above findings were suggestive of ectopia cordis, omphalocele, dysplastic kidneys with megabladder. Cardiac defects could not be adequately evaluated due decreased amniotic fluid.

Subsequently, MRI in T1, T2, T2 SPAIR and TSE in axial and coronal planes were done which confirmed the ectopia cordis, omphalocele, megabladder and additionally, skeletal deformities were revealed with scoliosis of the spine and malrotation of pelvis [Table/Fig-7-10].



[Table/Fig-7]: T1 coronal section showing the hypointense bladder with hyper-iso-intense liver superior to it and bowel adjacent to it.

DISCUSSION

Ectopia cordis is a rare congenital anomaly which occurs in 8 per million live births. The most common association is the Pentalogy of Cantrell (PC). Antenatal ultrasound is very accurate in diagnosing ectopia cordis as early as second trimester. The prognosis is usually poor.

Pentalogy of Cantrell consists of 5 defects in the developing fetus. The two major defects are the ectopia cordis and anterior abdominal wall defect. The other three defects are defect in the distal sternum, anterior diaphragm and the diaphragmatic pericardium. It was described by Cantrell in 1958 [1] and occurs sporadically with variable degrees of expression. Its incidence is 1 in 65000-200,000 with male is to female ratio 2:1. Males are more seriously affected than females [2].

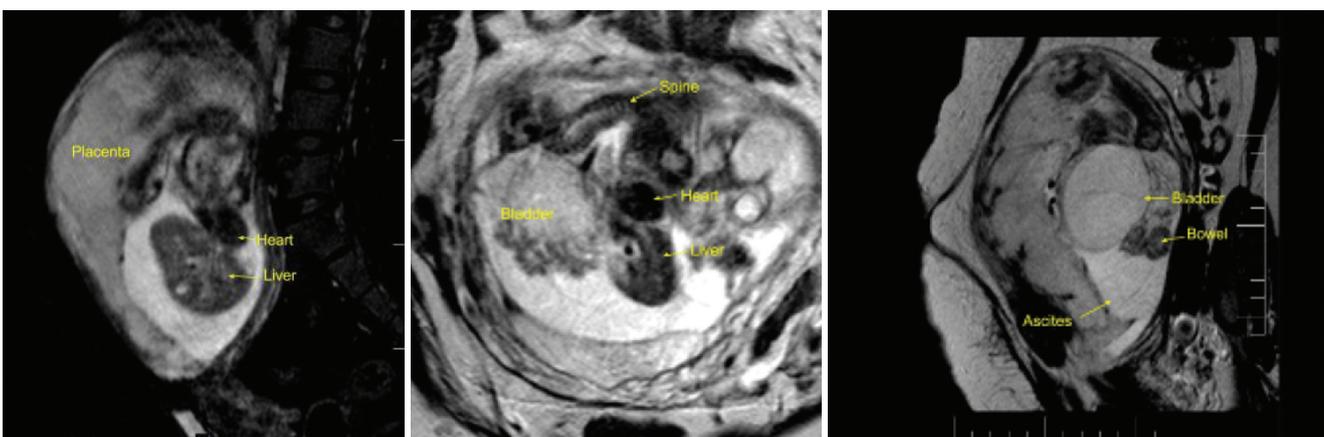
PC can be classified into 3 classes depending on the number of malformations:

- Diagnosis 'certain' if all 5 defects are present.
- Diagnosis probable if 4 defects (including intracardiac and ventral abdominal wall) are present.
- Diagnosis is incomplete if variable combinations of defects are present (always including a sternal abnormality) [3].

It has been hypothesized in literature that it occurs due to a developmental failure of a segment of the lateral mesoderm at around 14-18 days of embryonic life. As a consequence the transverse septum of the diaphragm does not develop and the ventromedial migration of the paired mesodermal folds fails to occur [4]. Martin et al., independently in their study mapped a gene to Xq25-q26.1 in patients with PC [5].

The typical finding in PC is detecting cardiac activity outside the chest and an anterior abdominal wall defect, ascites may occur due to compression of the contents of the chest and abdomen.

Ectopia cordis can be cervical, cervicothoracic, thoracic and thoracoabdominal in location [3]. Patients with thoracoabdominal ectopia cordis have better prognosis than others [6]. Cardiac anomalies associated with PC are atrial septal defect (53%), ventricular septal defect (100%), tetralogy of fallot (20%), tricuspid atresia, double outlet right ventricle, ventricular diverticulum (20%) [1].



[Table/Fig-8]: T2 SPAIR sequence showing extra-abdominal iso-intense liver and hypointense ectopia cordis with reduced thoracic circumference. Placenta seen anteriorly. **[Table/Fig-9]:** T2 coronal section showing acute kyphotic spinal deformity and omphalocele with membranes separating it from the slightly hypo intense amniotic fluid. **[Table/Fig-10]:** 10 MRI T2 coronal section with fetal ascites and thin membrane covering the abdominal contents.

Prognosis does not depend on the severity of the intracardiac defects, rather, It is the location of the EC along with the severity of abdominal & diaphragmatic defect and associated pulmonary hypoplasia that determines the prognosis [6].

Other associated non-cardiac anomalies are cranio-facial anomalies, hydrocephalus, anencephaly, chromosomal abnormalities such as trisomy 18, malrotation of the colon, cystic hygroma, dysplastic kidney and club foot, single umbilical artery, absent gall bladder, gastroschisis, eventration, diastasis recti [2,7]. The most commonly associated anomalies are the abdominal wall defects, usually an omphalocele or a gastroschisis with omphalocele being more common [6].

The most useful antenatal mode of investigation is ultrasound which is accurate in diagnosing this condition as early as mid-second trimester [6]. Additionally, a fetal MRI can be performed to confirm the diagnosis and to evaluate any additional anomalies, while karyotyping may give information on chromosomal abnormalities.

Management of PC is usually termination of pregnancy due to its poor prognosis. Postnatally, palliative surgery is performed. Prognosis depends on the severity of intracardiac and extra cardiac defects. Complex cases don't survive beyond few days of age while less severe cases with palliative treatment may live for longer.

A corrective surgery for ectopia cordis is performed by covering it with a skin flap and repositioning it into the chest. Repair of the omphalocele, diaphragmatic defect and sternal defects are done preferably as a one stage procedure. Some centers have got some success in managing the neonate with staged procedures [5].

When PC is diagnosed, a multidisciplinary team including an obstetrician, a neonatologist, a pediatric cardiologist, a pediatric surgeon, and a genetics specialist should inform the families about the prognosis of the disease. The option of termination of pregnancy should be always available for these patients [8].

CONCLUSION

Our case is most likely to be a case of Pentalogy of Cantrell (Thoraco-abdominal syndrome), as the two major components of PC, namely, ectopia cordis and omphalocele were demonstrated. Due to lack of follow up, autopsy of the neonate could not be performed and hence, other components of Pentalogy of Cantrell could not be established. Our case also had unusual associations like a megabladder, dysplastic kidneys, malrotated pelvis and spine deformity.

Detecting anomalies in the late second trimester and third trimester is relatively difficult by ultrasound, MRI gives additional information and helps in confirming the diagnosis.

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